

## HubExome Plus Panel - GeneSGKit®

CE-IVD



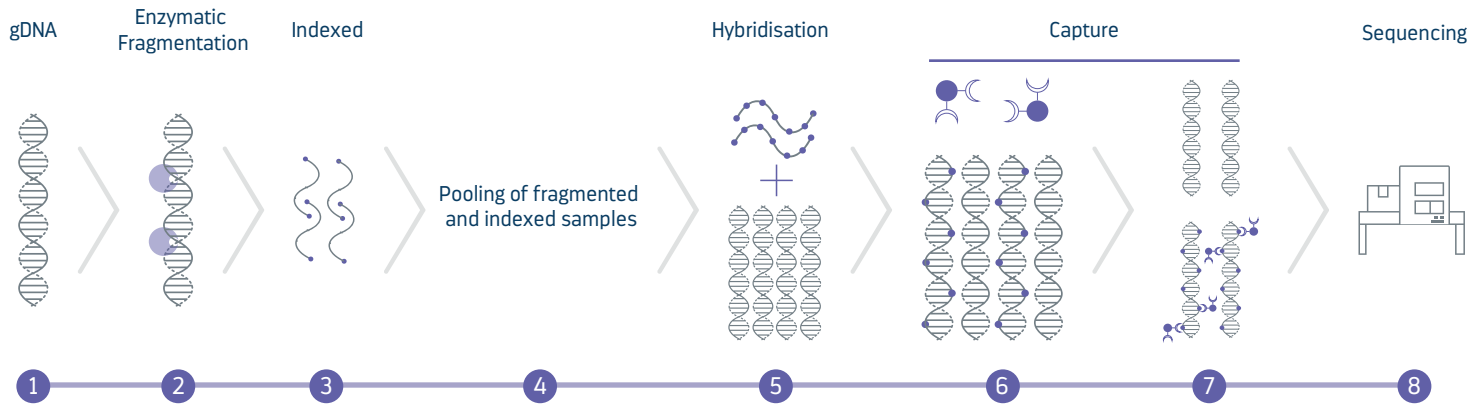
**Single kit for the study of all human pathologies from whole exome capture and mitochondrial DNA with CE-IVD marking.**

- ✓ **Single integrated solution on the market:**  
HubExome Plus Panel® includes reagents to process 16-48 samples with bioinformatic computation (secondary analysis) and visualization, prioritization and interpretation system included (tertiary analysis). The kit, secondary and tertiary analysis have CE-IVD marking.
- ✓ **Maximum coverage, sensitivity and specificity:**  
Captures all exonic regions of the genome (>19000 genes), flanking regions (+/- 20 bp) and mtDNA with a compact size of ~ 40 Mb.
- ✓ **Comprehensive study of all types of mutations:**  
Validated capture for the detection of SNVs, INDELs, large INDELs, MNVs, ALUs and CNVs.
- ✓ **Absolute traceability of samples:**  
Sample tracking system included in order to ensure a correct association between patient and result.
- ✓ **Speed:**  
Libraries are ready in just 12 hours from 100 ng of DNA.
- ✓ **Automation:**  
Possibility of automating the wet process with different commercial robots for generating libraries.
- ✓ **Complete bioinformatic analysis and results visualization with GeneSystems®:**  
The data is computed in less than 48 hours, scoring against the latest versions of more than 100 databases, both public and private.

**HubExome Plus Panel®** includes within the CE-IVD scope the analysis of the following pathologies:

- Bone Dysplasia and Collagen Disorders
- Cardiology
- Ectodermal Diseases
- Endocrinology
- Epilepsy
- Gastroenterological Diseases
- Hearing Loss
- Hematology
- Immunology
- Intellectual Disability and Autism Spectrum Disorder
- Metabolopathy
- Mitochondrial Diseases
- Neurology
- Ophthalmology
- Renal Diseases

# WORKFLOW de GeneSGKit®



## ABOUT US

Sistemas Genómicos is a pioneering company in the use of next-generation sequencing for genetic diagnosis. With more than 6,000 analyses per year, we are leaders in the sector, supported by years of experience and accredited and certified by the National Accreditation Body of Spain (ENAC) and the Spanish Association for Standardisation and Certification (AENOR).

## OUR OBJECTIVE

To capitalise on all our experience in analysis and diagnosis by using NGS to develop a series of tools that enable genetic diagnosis in situ in hospitals and research centres. We have achieved this by developing a comprehensive solution, the only one of its kind on the market, based on more than 35 GeneSGKits® for various diseases, all with CE-IVD marking and the GeneSystems®.



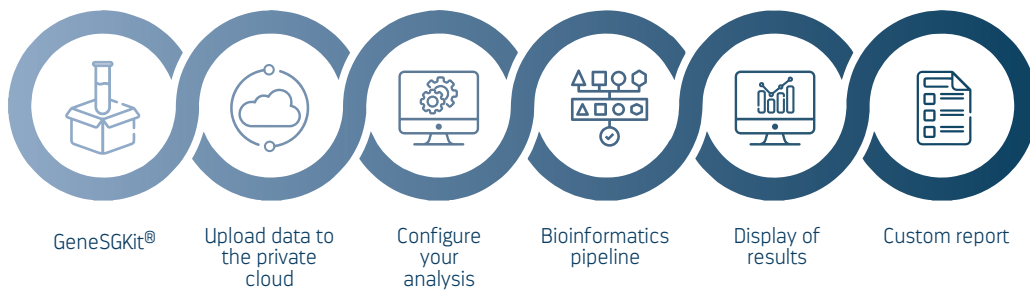
HubExome Plus Panel® provides high performance from only 100 ng of DNA. The design of the capture probes and bioinformatic pipeline guarantees high sensitivity and specificity for the detection of SNVs, indels, large indels, MNVs, ALUs and CNV screening with a recommended average coverage of 100x.



Sistemas Genómicos understands the vital importance of quality customer service and follow-up. This includes a personalised service that offers not only support over the telephone for all sorts of questions and concerns that may arise while using GeneSGKits® or the GeneSystems® analysis platforms, but also personalised training for the user who purchases the product. These training sessions will facilitate the set-up and roll-out of a genetic diagnostic service in situ at hospitals and laboratories using GeneSGKits®.

GeneSystems® is our bioinformatics tool for genetic diagnosis and research. It is a platform registered as a medical device with CE marking for diagnostic use *in vitro*, to process information from massive DNA sequencing, detect variants and help genetic diagnosis. Developed in a **Cloud Computing** environment, it can provide access your data from any computer device, at any time and place, without consuming the user's own resources.

GeneSystems® is designed to provide a solution to the different stages of the NGS data analysis step:

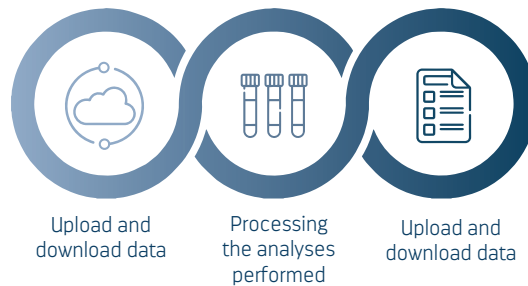


## FEATURES OF GENESYSTEMS®

Integral infrastructure **bioinformatic analysis** for the data analysis from NGS platforms.

**Automation** of the entire data analysis process.

**Cloud Computing platform:** access from any device with an internet connection.



It was developed by a multidisciplinary team of geneticists, bioinformaticians and engineers, and has been validated by over 200 companies, including hospitals, laboratories, research centres, universities and pharmaceutical companies. The platform has been independently audited and detects **SNVs, INDELS, large INDELS, MNVs, ALUs, CNVs and SVs**.

### ANALYSIS OF SEQUENCING DATA WITH GENEBYTES

#### In the laboratory routine

- Cloud-based platform: no software installation required.
- Upload and download results quickly and easily.
- Bioinformatic analysis of results from raw data, BAM and VCF.
- Exhaustive report of the computation process.
- Resource optimisation: support for preparing pools of non-equimolar libraries prior to sequencing.
- View the results of computations in the corresponding application.

#### In research

- Gene, exome and genome panels: view and interact with the results obtained through NGS.
- Annotation of variants compared to the major public and private databases.
- Flexible filtering.
- Simultaneous analysis of samples.

#### In diagnosis

- Flexible viewing and prioritisation of the variants obtained.
- Identification of point variants, Indels, large Indels and structural variants (balanced and unbalanced).
- Simultaneous analysis of samples: optimised diagnostic analysis time.
- Full annotation for variant classification:
  - DBNLVar (Sistemas Genómicos' own database).
  - Public and private reference databases.
  - *In silico* predictors.
- Integrated IGV for a graphic analysis of the genomic environment.
- Customisable reports of genetic results.
- Generation of virtual panels to prioritise variants according to HPO terminology.
- Automatic classification of variants based on ACMG guidelines.
- Prioritisation of results using combined algorithms
  - Phenotype (HPO)
  - Target tissue expression (GTEx)
  - Pathogenicity (ACMG guidelines)
- Manages the primary databases in the sector (DBNLVar, Ensembl, 1000Genomes, HGMD-Pro, ClinVar, etc.)
  - Pathogenicity indicators with links to scientific papers.

# CATALOGUE - HubExome Plus Panel - GeneSGKit®

## Technical data

GeneSGKit®

Size (MB)

HubExome Plus Panel - GeneSGKit®

40

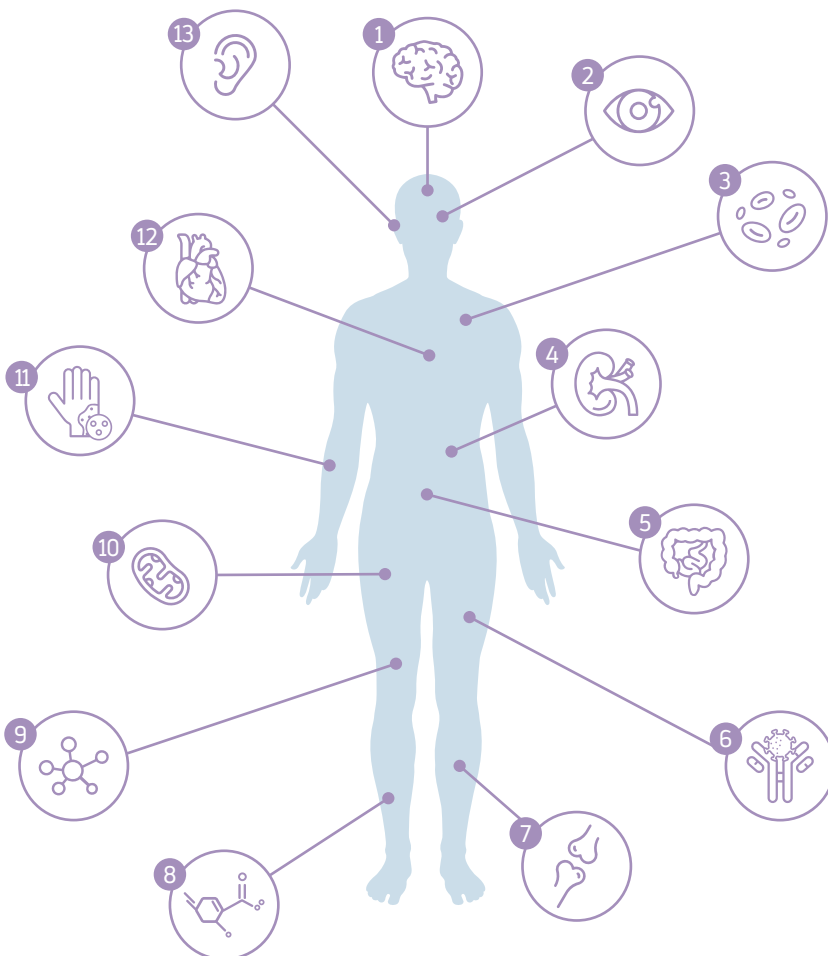
## References

HubExome Plus Panel® 16 reactions Ref.: LV4612 (CE-IVD)

HubExome Plus Panel® 48 reactions Ref.: LV4613 (CE-IVD)



Virtual gene panels design has been carried out following the most up-to-date international guidelines and scientific recommendations. In addition, **full coverage** of exons and +/-20 bp of intronic and UTR regions is ensured for all of included genes.



- 1 Neurology | 806 genes  
Epilepsy | 523 genes  
Intellectual Disability and Autism Spectrum Disorder | 1564 genes
- 2 Ophthalmology | 508 genes
- 3 Hematology | 153 genes
- 4 Renal Diseases | 339 genes
- 5 Gastroenterological Diseases | 153 genes
- 6 Immunology | 458 genes
- 7 Bone Dysplasia and Collagen Disorders | 608 genes
- 8 Metabolopathy | 400 genes
- 9 Endocrinology | 253 genes
- 10 Mitochondrial Diseases | 384 genes
- 11 Ectodermal Diseases | 323 genes
- 12 Cardiology | 608 genes
- 13 Hearing Loss | 179 genes